The Role of Single Nucleotide Polymorphisms (SNPs) in Understanding Complex Disorders and Pharmacogenomics

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Abstract

Introduction: In the last two years, there has been an increasing interest in single nucleotide polymorphisms (SNPs). They have been hailed as the most common polymorphism found in the human genome and are believed to be responsible for 90% of all inter-individual variation. Efforts are now directed at the large-scale identification and archiving of SNPs in the human genome. Not only are they useful markers for population divergence studies, SNPs can be utilised as markers in studies of complex diseases and pharmacogenomics. Methods: Traditional methods for identifying SNPs, as well as methods for large-scale detection and genotyping of SNPs currently being developed, are briefly discussed in this review. Such developments will facilitate and enhance the process of identifying and characterising genes and their functions. Results: The utility of SNPs in identifying genes contributing to pharmacogenetic variation and increased risk of a complex disease is discussed. The role of SNPs in influencing drug response in different individuals is also presented. Conclusions: In helping to unravel the genetic basis of complex diseases and inter-individual variation in drug response, SNPs will catalyse the transition into a new age of medicine in which medical care is tailored to the individual’s genetic profile.


Key words: Complex diseases, Drug response, Gene discovery, Pharmacogenetics, Single nucleotide polymorphism

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